



DNAH5 gene

dynein axonemal heavy chain 5

Normal Function

The *DNAH5* gene provides instructions for making a protein that is part of a group (complex) of proteins called dynein. This complex functions within cell structures called cilia. Cilia are microscopic, finger-like projections that stick out from the surface of cells. Coordinated back and forth movement of cilia can move the cell or the fluid surrounding the cell. Dynein produces the force needed for cilia to move.

Within the core of cilia (the axoneme), dynein complexes are part of structures known as inner dynein arms (IDAs) or outer dynein arms (ODAs) depending on their location. Coordinated movement of the dynein arms causes the entire axoneme to bend back and forth. IDAs and ODAs have different combinations of protein components (subunits) that are classified by weight as heavy, intermediate, or light chains. The *DNAH5* gene provides instructions for making heavy chain 5, which is found in ODAs. Other subunits are produced from different genes.

Health Conditions Related to Genetic Changes

heterotaxy syndrome

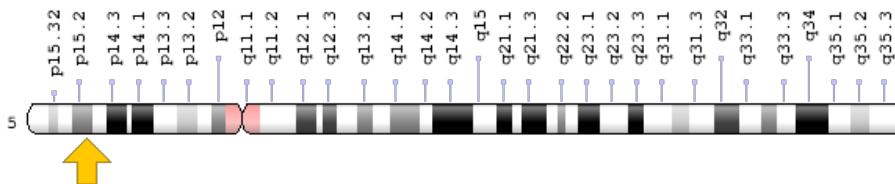
primary ciliary dyskinesia

More than 80 mutations in the *DNAH5* gene have been found to cause primary ciliary dyskinesia, which is a condition characterized by respiratory tract infections, abnormal organ placement, and an inability to have children (infertility). *DNAH5* gene mutations result in an absent or abnormal heavy chain 5. Without a normal version of this subunit, the ODAs cannot form properly and may be shortened or absent. As a result, cilia cannot produce the force needed to bend back and forth. Defective cilia lead to the features of primary ciliary dyskinesia.

Chromosomal Location

Cytogenetic Location: 5p15.2, which is the short (p) arm of chromosome 5 at position 15.2

Molecular Location: base pairs 13,690,328 to 14,011,829 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- axonemal beta dynein heavy chain 5
- CILD3
- ciliary dynein heavy chain 5
- DNAHC5
- DYH5_HUMAN
- dynein heavy chain 5, axonemal
- dynein, axonemal, heavy chain 5
- dynein, axonemal, heavy polypeptide 5
- FLJ46759
- HL1
- KIAA1603

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Cilia and Flagella: Structure and Movement
<https://www.ncbi.nlm.nih.gov/books/NBK21698/>

GeneReviews

- Primary Ciliary Dyskinesia
<https://www.ncbi.nlm.nih.gov/books/NBK1122>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28DNAH5%5BTIAB%5D%29+OR+%28DNAHC5%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- DYNEIN, AXONEMAL, HEAVY CHAIN 5
<http://omim.org/entry/603335>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=DNAH5%5Bgene%5D>
- HGNC Gene Family: Dyneins, axonemal
<http://www.genenames.org/cgi-bin/genefamilies/set/536>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2950
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1767>
- UniProt
<http://www.uniprot.org/uniprot/Q8TE73>

Sources for This Summary

- OMIM: DYNEIN, AXONEMAL, HEAVY CHAIN 5
<http://omim.org/entry/603335>
- Djakow J, Svobodová T, Hrach K, Uhlík J, Cinek O, Pohunek P. Effectiveness of sequencing selected exons of DNAH5 and DNAI1 in diagnosis of primary ciliary dyskinesia. *Pediatr Pulmonol*. 2012 Sep;47(9):864-75. doi: 10.1002/ppul.22520. Epub 2012 Mar 13.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/22416021>
- Escudier E, Duquesnoy P, Papon JF, Amselem S. Ciliary defects and genetics of primary ciliary dyskinesia. *Paediatr Respir Rev*. 2009 Jun;10(2):51-4. doi: 10.1016/j.prrv.2009.02.001. Epub 2009 Apr 18. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19410201>

- Failly M, Bartoloni L, Letourneau A, Munoz A, Falconnet E, Rossier C, de Santi MM, Santamaria F, Sacco O, DeLozier-Blanchet CD, Lazor R, Blouin JL. Mutations in DNAH5 account for only 15% of a non-preselected cohort of patients with primary ciliary dyskinesia. *J Med Genet.* 2009 Apr;46(4):281-6. doi: 10.1136/jmg.2008.061176.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19357118>
- GeneReview: Primary Ciliary Dyskinesia
<https://www.ncbi.nlm.nih.gov/books/NBK1122>
- Hornef N, Olbrich H, Horvath J, Zariwala MA, Fliegauf M, Loges NT, Wildhaber J, Noone PG, Kennedy M, Antonarakis SE, Blouin JL, Bartoloni L, Nüsslein T, Ahrens P, Griese M, Kuhl H, Sudbrak R, Knowles MR, Reinhardt R, Omran H. DNAH5 mutations are a common cause of primary ciliary dyskinesia with outer dynein arm defects. *Am J Respir Crit Care Med.* 2006 Jul 15; 174(2):120-6. Epub 2006 Apr 20.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16627867>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2662904/>
- Leigh MW, Pittman JE, Carson JL, Ferkol TW, Dell SD, Davis SD, Knowles MR, Zariwala MA. Clinical and genetic aspects of primary ciliary dyskinesia/Kartagener syndrome. *Genet Med.* 2009 Jul;11(7):473-87. doi: 10.1097/GIM.0b013e3181a53562. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/19606528>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3739704/>
- Olbrich H, Häffner K, Kispert A, Völkel A, Volz A, Sasmaz G, Reinhardt R, Hennig S, Lehrach H, Konietzko N, Zariwala M, Noone PG, Knowles M, Mitchison HM, Meeks M, Chung EM, Hildebrandt F, Sudbrak R, Omran H. Mutations in DNAH5 cause primary ciliary dyskinesia and randomization of left-right asymmetry. *Nat Genet.* 2002 Feb;30(2):143-4. Epub 2002 Jan 14.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/11788826>

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